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 NEWS 10  Jun 10  MEDLINE Reload
 NEWS 11  Jun 10  PCTFULL has been reloaded
 NEWS 12  Jul 02  FOREGE no longer contains STANDARDS file segment
 NEWS 13  Jul 22  USAN to be reloaded July 28, 2002;
                  saved answer sets no longer valid
 NEWS 14  Jul 29  Enhanced polymer searching in REGISTRY
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L2          111 L1 (P) POLYMORPH? SITE#

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SO   Genetics, (April, 2002) Vol. 160, No. 4, pp. 1619-1630. print.
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AU   Hattori, Nobuaki; Kitagawa, Kazuaki; Takumi, Shigeo; Nakamura, Chiharu
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TI Diagnostic method and kit for determining Kell blood group genotype

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PA New York Blood Center, Inc., USA
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| | PATENT NO. | KIND | DATE | APPLICATION NO. | DATE |
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| PI | WO 9615268 | A1 | 19960523 | WO 1995-US14684 | 19951109 |
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CS MRC Immunodeficiency Research Group, Royal Free Hospital of Medicine, London, NW3 2PF, UK
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L3 ANSWER 19 OF 40 BIOSIS COPYRIGHT 2002 BIOLOGICAL ABSTRACTS INC.DUPLICATE
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AB In this work, we explored the existence of genetic variants within the apolipoprotein E gene transcriptional regulatory region, using a denaturing gradient gel electrophoresis screening of a region comprising nucleotides -1017 to +406. Upon a population study, three new **polymorphic sites** (-491, -427 and -219) and two mutations were found. Functional effects of the polymorphisms, assayed by transient transfection and electrophoretic mobility shift assays in a human hepatoma cell line, showed that polymorphisms at sites -491 and -219 of the APOE promoter produce variations in the transcriptional activity of the gene, most probably through **differential** binding of nuclear proteins.

L3 ANSWER 22 OF 40 CAPLUS COPYRIGHT 2002 ACS
AB The invention provides a diagnostic method of detg. Kell genotype by the identification of the mol. basis of a Kell polymorphism. Specifically, the invention provides a method for detg. K1/K2 genotype with great accuracy, overcoming problems assocd. with traditional serol. typing methods. The diagnostic method of the invention preferably employs amplification of K1/K2 nucleic acid sequences, and optionally employs **differential** cleavage of K1-K2-specific nucleic acid sequences by a restriction enzyme. Also provided are nucleic acid oligomers useful as probes or primers for the method of the invention. Furthermore, diagnostic kits for the detn. of Kell genotype are provided. Thus, the complete sequence of the KEL gene from a human placental genomic DNA library indicated that the gene is organized into 19 exons. PCR cloning and sequencing indicated the mol. basis for the K1/K2, K6/K7, K10/K(-10), and K3/K4/K21 polymorphisms: the K1/K2 polymorphism occurs in exon 6 resulting in a Thr192.fwdarw.Met substitution in the Kell protein; the K6/K7 polymorphism is in exon 17 resulting in a Leu597.fwdarw.Pro substitution; a point exon 13 results in Glu496.fwdarw.Val for the K10/K(-10) polymorphism; and the K3/K4/K21 polymorphism is in exon 8 with an Arg281.fwdarw.Trp.fwdarw.Gln substitution. Each of these **polymorphic sites** cleavable by specific restriction endonucleases (BsmI for K1/K2, MnlI or DdeI for K6/K7, AccI for K10/K(-10), and NlaIII or PvuII for K3/K4/K21) or amplifiable by allele-specific PCR primers. In addn., alloimmunization can be detd. with
Kell antigen peptide probes reactive to anti-Kell antibodies.

L3 ANSWER 24 OF 40 CAPLUS COPYRIGHT 2002 ACS
AB For rapid individual anal. of patients with X-linked agammaglobulinemia (XLA) and for disease carriers, an exon-scanning approach has been developed using a single PCR to amplify all exons and the putative promoter region with a single annealing temp. and with the same PCR conditions for all amplified segments. This oligonucleotide primer set has now been improved to amplify individually all Bruton's tyrosine kinase
(BTK) exons, to **enhance** the yield and purity of the PCR products, to avoid described **polymorphic sites** in the amplified segments, and to solve inconsistencies in the intronic sequences
generated independently by several groups. Novel missense mutations I429N
and M477R were identified. Thus, a rapid and efficient system for mutation detection of the BTK gene is presented which allows an improved mol. diagnosis of individual patients with XLA, including presymptomatic testing for a putative gene defect and **differential** diagnosis of

human primary immunodeficiencies with a reduced no. of B-cells. A non-random distribution of amino acid substitutions along the enzyme domains has been demonstrated using the available BTK mutation pattern in patients with XLA.

L3 ANSWER 27 OF 40 BIOSIS COPYRIGHT 2002 BIOLOGICAL ABSTRACTS
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AB Several repetitive elements have been associated with and identified in the surrounding region and within the human c-H-ras1 gene. The polymorphism exhibited by these sites provides valuable information regarding the function and structure of the H-ras gene. We have investigated two such **polymorphic sites**: i) a hexanucleotide microsatellite region (HRMS) within intron 1 of H-ras and ii) the VTR region at the 3' end of the gene. Comparison between normal and tumor tissues from 25 samples of bladder cancer revealed that 5 samples (20%) exhibited LOH at these **polymorphic sites** indicating that an allelic loss had occurred at the H-ras locus. Furthermore, instability was detected in 4 cases at the hexanucleotide locus, while the VTR region was found unaffected. The two **polymorphic sites** are in a strong linkage disequilibrium in normal tissues, while in tumor tissues with genomic instability this linkage is altered, possibly leading to **differential** regulation of the H-ras. Also a previously reported allele at the HRMS locus was found to behave in a manner that preserves the linkage between the two **polymorphic sites** in normal tissues.

=> s PCR amplification (S) stem loop primers
L4 0 PCR AMPLIFICATION (S) STEM LOOP PRIMERS

=> s PCR amplification (S) hairpin primers
L5 0 PCR AMPLIFICATION (S) HAIRPIN PRIMERS

=> s PCR amplification and hairpin primers
L6 1 PCR AMPLIFICATION AND HAIRPIN PRIMERS

=> d l6

L6 ANSWER 1 OF 1 CAPLUS COPYRIGHT 2002 ACS
AN 2002:555680 CAPLUS
TI Fluorescent dye-labeled primers containing base or sugar analogs for improved sensitivity and control of artifacts in nucleic acid amplification
IN Nazarenko, Irina; Rashtchian, Ayoub; Solus, Joseph; Pires, Richard M.; Darfler, Marlene; Gebeyehu, Gulilat; Astatke, Mekbib
PA Invitrogen Corporation, USA
SO PCT Int. Appl., 307 pp.
CODEN: PIXXD2
DT Patent
LA English
FAN.CNT 1

| | PATENT NO. | KIND | DATE | APPLICATION NO. | DATE |
|---|---|---|---|---|---|
| PI | WO 2002057479 | A2 | 20020725 | WO 2001-US50460 | 20011227 |

W: AE, AG, AL, AM, AT, AU, AZ, BA, BB, BG, BR, BY, BZ, CA, CH, CN, CO, CR, CU, CZ, DE, DK, DM, DZ, EC, EE, ES, FI, GB, GD, GE, GH, GM, HR, HU, ID, IL, IN, IS, JP, KE, KG, KP, KR, KZ, LC, LK, LR, LS, LT, LU, LV, MA, MD, MG, MK, MN, MW, MX, MZ, NO, NZ, OM, PH, PL, PT, RO, RU, SD, SE, SG, SI, SK, SL, TJ, TM, TN, TR, TT, TZ, UA, UG, UZ, VN, YU, ZA, ZM, ZW, AM, AZ, BY, KG, KZ, MD, RU, TJ, TM

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        RW: GH, GM, KE, LS, MW, MZ, SD, SL, SZ, TZ, UG, ZM, ZW, AT, BE, CH,
            CY, DE, DK, ES, FI, FR, GB, GR, IE, IT, LU, MC, NL, PT, SE, TR,
            BF, BJ, CF, CG, CI, CM, GA, GN, GQ, GW, ML, MR, NE, SN, TD, TG
PRAI US 2000-748146    A     20001227
     US 2001-330468P   P     20011023


=>

---Logging off of STN---


=>
Executing the logoff script...

=> LOG Y

COST IN U.S. DOLLARS                                SINCE FILE      TOTAL
                                                       ENTRY    SESSION
FULL ESTIMATED COST                                   114.59     114.80

DISCOUNT AMOUNTS (FOR QUALIFYING ACCOUNTS)          SINCE FILE      TOTAL
                                                       ENTRY    SESSION
CA SUBSCRIBER PRICE                                    -1.24      -1.24

STN INTERNATIONAL LOGOFF AT 11:31:07 ON 13 AUG 2002
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